

## Opis choroby \*

### Definicja

A rare genetic neurological disorder characterized by hypotonia, delayed motor development, dyskinesia of the limbs, intellectual disability with impaired speech development, seizures, autistic features, stereotypic movements, and sleep disturbance. Onset of symptoms is in infancy. Bilateral abnormalities in the putamen on brain MRI have been reported in some patients.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

468620

#### Kod OMIM

617171

#### Kod ICD10

G93.8

#### Kod ICD11

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### \*Źródło

orphanet